



## ERAP1 gene

endoplasmic reticulum aminopeptidase 1

### Normal Function

The *ERAP1* gene (also known as *ERAAP* and *ARTS1*) provides instructions for making a protein called endoplasmic reticulum aminopeptidase 1. As its name suggests, this protein is active in a cellular structure called the endoplasmic reticulum, which is involved in protein processing and transport. This protein is an aminopeptidase, which is an enzyme that cuts (cleaves) other proteins into smaller fragments called peptides.

Endoplasmic reticulum aminopeptidase 1 has two major functions, both of which are important for normal immune system function. First, endoplasmic reticulum aminopeptidase 1 cleaves several proteins called cytokine receptors on the surface of cells. Cleaving these receptors reduces their ability to transmit chemical signals into the cell, which affects the process of inflammation.

Second, endoplasmic reticulum aminopeptidase 1 cleaves many types of proteins into small peptides that can be recognized by the immune system. These peptides are exported to the cell surface, where they attach to major histocompatibility complex (MHC) class I proteins. MHC class I proteins display the peptides to the immune system. If the immune system recognizes the peptides as foreign (such as viral or bacterial peptides), it responds by triggering the infected cell to self-destruct.

### Health Conditions Related to Genetic Changes

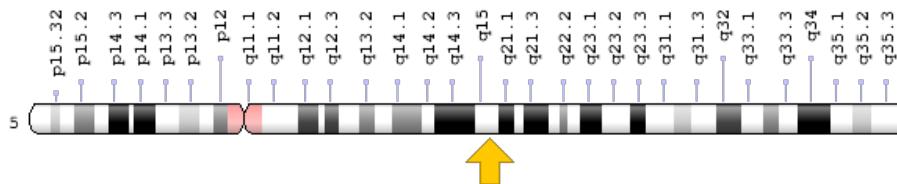
#### ankylosing spondylitis

Several variations (polymorphisms) in the *ERAP1* gene have been found to influence the risk of ankylosing spondylitis. Each of these variations changes a single protein building block (amino acid) in endoplasmic reticulum aminopeptidase 1. Little is known about the effects of these variations, although researchers believe that changes in the protein's structure could alter either of its two major functions. It is unclear how these changes contribute to a person's risk of ankylosing spondylitis. Other genetic and environmental factors, many of which are unknown, also affect the chance of developing this condition.

## Chromosomal Location

Cytogenetic Location: 5q15, which is the long (q) arm of chromosome 5 at position 15

Molecular Location: base pairs 96,759,245 to 96,935,983 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- A-LAP
- adipocyte-derived leucine aminopeptidase
- ALAP
- aminopeptidase PILS
- aminopeptidase regulator of TNFR1 shedding
- APPILS
- ARTS-1
- ARTS1
- ERAAP
- ERAAP1
- ERAP1\_HUMAN
- KIAA0525
- PILS-AP
- PILSAP
- puromycin-insensitive leucyl-specific aminopeptidase
- type 1 tumor necrosis factor receptor shedding aminopeptidase regulator

## **Additional Information & Resources**

### Educational Resources

- Immunobiology (fifth edition, 2001): The Major Histocompatibility Complex and Its Functions  
<https://www.ncbi.nlm.nih.gov/books/NBK27156/>

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ERAP1%5BTIAB%5D%29+OR+%28endoplasmic+reticulum+aminopeptidase+1%5BTIAB%5D%29%29+OR+%28ARTS-1%5BTIAB%5D%29+OR+%28ARTS1%5BTIAB%5D%29+OR+%28ERAAP%5BTIAB%5D%29+OR+%28PILS-AP%5BTIAB%5D%29%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

### OMIM

- ENDOPLASMIC RETICULUM AMINOPEPTIDASE 1  
<http://omim.org/entry/606832>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_ERAP1.html](http://atlasgeneticsoncology.org/Genes/GC_ERAP1.html)
- HGNC Gene Family: Aminopeptidases  
<http://www.genenames.org/cgi-bin/genefamilies/set/104>
- HGNC Gene Family: Minor histocompatibility antigens  
<http://www.genenames.org/cgi-bin/genefamilies/set/870>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=18173](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=18173)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/51752>
- UniProt  
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Reprinted from Genetics Home Reference:

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Reviewed: February 2009

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications  
U.S. National Library of Medicine  
National Institutes of Health  
Department of Health & Human Services